MedGen: A Medical Genetics Portal

A public web portal providing access to medical genetics information available from NCBI

https://www.ncbi.nlm.nih.gov/medgen

National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

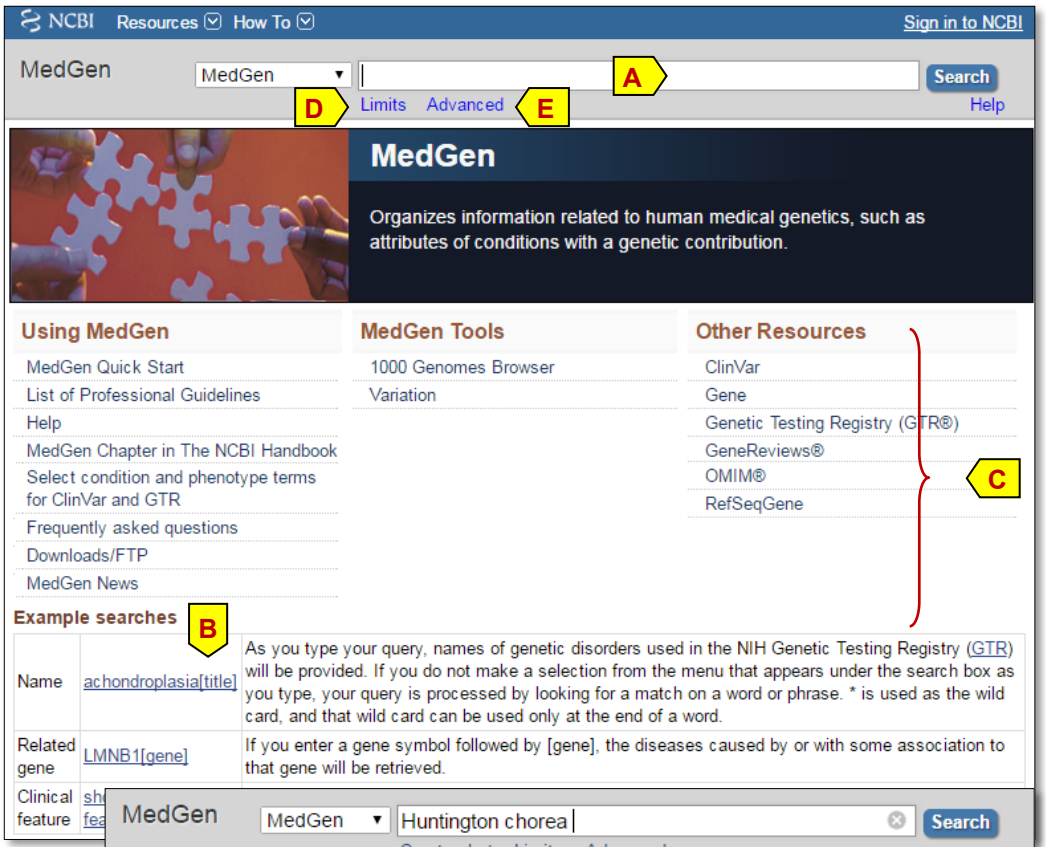
Scope and Access

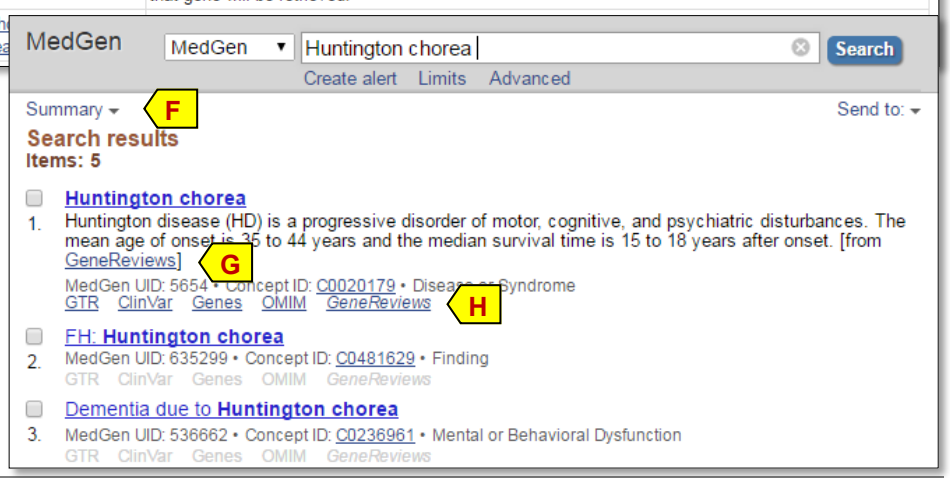
MedGen provides up to date information about human disorders and other phenotypes with a genetic component. This database integrates information from multiple sources, such as OMIM, SNOMED CT, and Human Phenotype Ontology, and thus conveys community standards for terms, definitions, descriptions, and identifiers. The content is augmented by the inclusion of professional guidelines curated by staff of the Genetic Testing Registry (GTR), as well as links to publications related to disorders, clinical studies and systematic reviews based on PubMed's Clinical Queries. Data in MedGen are acquired both programmatically and via curation, depending on the type of information and the sources for that information. The first step in organizing the information is to establish a concept that defines a disorder or phenotype, classify that concept by type, and then assign that concept a stable unique identifier. With thatframework established, data are connected to that concept.

MedGen is integrated with other biological, clinical and genetic testing resources at NCBI, e.g. the Gene database, the Genetic Testing Registry (GTR), ClinVar, GeneReviews, and Medical Genetics Summaries on NCBI's bookshelf. Data from MedGen can be searched on the web through its homepage (https://www.ncbi.nlm.nih.gov/medgen) or accessed programmatically via E-utilities. MedGen provides links to many resources to facilitate discovery. Terms and their relationships are available for download from MedGen's ftp site (ftp://ftp.ncbi.nlm.nih.gov/pub/medgen/).

Searching in MedGen

Searching for information in MedGen can be done by typing desired query terms in the search box followed by clicking the “Search” button (**A**). For more specific data retrieval, field -limited terms connected with appropriate Boolean operators (AND, OR, NOT) are strongly recommended. The “Example searches” section (**B**) provides a few representative examples of field-limited terms. Clicking an underlined example term retrieves a set of current records. Links to documents on MedGen and relevant tools/resources from NCBI are listed in the middle section (**C**). The “Limits” (**D**) links to the Limits page, which provides access to a set of preset fields to allow quick retrieval of records with certain characteristics. Advanced” (**E**) links to a query construction page, where sophisticated queries can be constructed using Boolean operator connected terms limited to selected field for more specific data interrogation.





Search results are displayed in the summary format (**F**). The information contained within a record can be gauged by the data source (**G**) and the number of connections to relevant records in other resources (**H**). Clicking the hyperlinked title of a record opens it in full report format.

Information Provided by a MedGen Record

Clicking the title of a search retrieved record opens it in the "full report" display format (**A**). This report is organized into clearly marked sections, each addressing a specific aspect of the disease. The "Table of contents" (**B**) provides a quick way to navigate to sections of interest. Contents under these sections will vary for individual records. MedGen records are integrated with other public resources and available information is organized as links under the "Table of contents." Here genetic tests for the disease are collected in the "Genetic Testing Registry" section (**C**). Related records from other NCBI resources are in the "Related information" section at the end of the right-hand column (not shown). For Huntington's Chorea:

- The disease synonyms, SNOMED IDs, and gene information are given at the top (**D**).
- Phenotypic description of the disease are presented in the "Disease characteristics," "Additional descriptions," and "Clinical features" section (**E**).
- Two sets of hierarchical terms for the disease are listed under the "Term hierarchy" section (**F**).
- Testing and treatment guidelines and updated information from recent clinical studies retrieved using the PubMed Clinical Queries system are displayed in their own sections (**G**, collapsed).

Entries under "Clinical feature" allow searching by specific clinical features using controlled vocabularies. For example, clicking the arrow in front of "Abnormality of the nervous system" (**H**) reveals the controlled term Chorea. Clicking the term (**I**) opens a popup with more details on the term (from HPO). Clicking "Search on this feature" (**J**) retrieves other conditions (MedGen records) with this clinical feature, highlighting the fact that a given clinical feature is present in different conditions.

Help Documents and MedGen Specific Help

MedGen online help documentation and handbook contain additional details:
http://www.ncbi.nlm.nih.gov/medgen/docs/help/ and http://www.ncbi.nlm.nih.gov/books/NBK159970/
Questions and comments should be addressed to: medgen_help@nih.gov